Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

July 2017

FEATURED DISORDER

Phenylketonuria

Phenylketonuria (PKU) was the first disorder to be screened on blood spot filter paper. In 1963, Dr. Robert Guthrie developed a revolutionary new way to test for PKU by collecting blood from a baby's heel onto a special filter paper. Dr. Guthrie's test was so successful states began implementing laws to make sure every baby was tested for PKU shortly after birth. This is why many people still call the blood spot screen the "PKU test" despite the fact that there have been many more disorders added to the test over the years.

About one in every 10,000 babies is born with PKU. At birth, these babies look and act completely normal, but babies with this genetic disorder cannot break down an amino acid called phenylalanine. Phenylalanine is found in all foods that contain protein and also in artificial sweeteners. If a baby with PKU is not placed on a special diet soon after birth, phenylalanine will build up in the baby's body causing brain damage and possibly death. Within a few months of age a baby with untreated PKU will begin to show symptoms such as irritability, seizures, eczema, musty body odor, and developmental delays. However, if PKU is detected early and treatment is initiated soon after birth, these symptoms can be prevented and babies can grow up to lead normal, healthy lives.

Each year Missouri's newborn screening program detects about five to 10 babies who have PKU. Timely and accurate blood spot collection at birthing hospitals throughout the state helps to ensure these babies can be placed on treatment early, giving them the opportunity to walk, talk, run, and play just like any other child.

What's New?

Welcome to **Behind the Screens**, a newsletter featuring information for you: the frontline, first-point-of-contact, vital component of the newborn screening process.

Newborn Screening is a series of tests performed on newborns shortly after birth to protect them from the dangerous effects of disorders that otherwise may not be detected for days, months, or even years.

In Missouri, babies are required by law to receive a bloodspot screening, hearing screening, and critical congenital heart disease (CCHD) screening. Newborn screening detects over 70 different disorders including hearing loss and CCHDs. The screening tests are very efficient and provide newborns with the best opportunity for early intervention and treatment. The incidence of bloodspot and hearing screening combined is one affected baby in every 250 births.

Did You Know?

Failure to receive follow-up services diminishes the benefit of newborn hearing screening.

Congenital hearing loss affects one to three of every 1,000 newborns and is referred to as a neuro-developmental emergency.

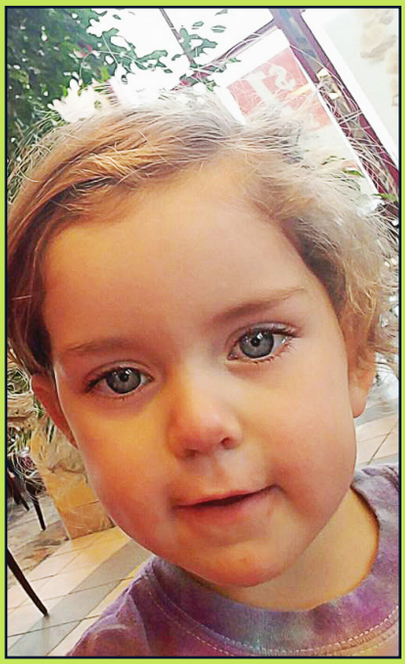
When left undetected, hearing loss can negatively impact children through delays in speech, language, social and emotional development. The Missouri Department of Health and Senior Services' Newborn Hearing Screening Program strives to ensure all deaf and hard-of-hearing infants are identified early and receive intervention services.

Hospital personnel assist in this process by screening infants for hearing loss prior to discharge, consistently reporting screening results to the DHSS and assisting parents to schedule follow-up services such as rescreening or audiologic evaluation.

Without prompt follow-up rescreening or audiologic evaluation, deaf and hard-of-hearing infants are at risk for developmental delays.

PATIENT SPOTLIGHT — *Georgia*

Georgia is a Missouri two-year-old with Pompe Disease. Thanks to the timely collection, transport and testing of her newborn screening sample, she was diagnosed and treated early enough to give her the quality of life which she enjoys today.



"It is a moment that not too many parents give a second thought. A tiny prick on the bottom of the foot. A nurse reassures you that they just need a few drops of blood for a newborn screen, and then it is done. Most parents never give it a second thought because they never hear another thing about it. Until you do. Then you are faced with questions, concerns and fears the like of which you have never experienced."

—Kari, Georgia's mother

Tech Tips

- Report hearing screening results no later than seven days from the date of screening.
- The critical congenital heart disease (CCHD) reporting screen now appears in MoEVR. You will be notified when data entry access is available.
- Accurate date and time of blood spot collection is crucial. This data cannot be amended once the specimen is received by the state public health laboratory.

Thank you for your contribution to ensuring the best possible start for Missouri newborns

Missouri Department of Health and Senior Services

Bureau of Genetics and Healthy Childhood—Newborn Blood Spot, Hearing, and CCHD Programs

573-751-6266 • 800-877-6246

Missouri State Newborn Screening Laboratory

573-751-2662

www.health.mo.gov/newbornscreening